

Claims

1 A method for the diagnosis of a polymorphism in a PDH E1 β gene in a human, which method comprises determining the sequence of the nucleic acid of the human at one or more of positions 457, 1191, 1198 and 1342 in the PDH E1 β gene as defined by the positions in
5 SEQ ID NO: 1; and determining the status of the human by reference to polymorphism in the PDH E1 β gene.

2 A method according to claim 1 in which the polymorphisms are further defined as:

Position	Polymorphism	Region
457	A/G	Coding, silent Gly
1191	A/C	3'UTR
1198	C/T	3'UTR
1342	C/A	3'UTR

10 3 A method according to claim 2 which comprises diagnosis of any one of the following haplotypes:

- (a) 1191C 1198C 1342A;
- (b) 1191A 1198C 1342C; or
- (c) 1191C 1198T 1342A.

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4 An isolated nucleic acid comprising the nucleic acid of SEQ ID NO: 1 with C at position 1191 as defined by the position in SEQ ID NO: 1; or a complementary strand thereof or an antisense sequence thereto or a fragment thereof of at least 20 bases comprising C at position 1191.

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5 An allele specific primer capable of detecting a PDH E1 β gene polymorphism at one or more of positions 457, 1191, 1198 and 1342 in the PDH E1 β gene as defined by the positions in SEQ ID NO: 1.

6 An allele-specific oligonucleotide probe capable of detecting a PDH E1 β gene polymorphism at one or more of positions 457, 1191, 1198 and 1342 in the PDH E1 β gene as defined by the positions in SEQ ID NO: 1.

5 7 Use of any polymorphism as defined in claim 2 as a genetic marker in a linkage study.

8 A method of treating a human in need of treatment with a PDH drug in which the method comprises:

10 i) diagnosis of a polymorphism in the PDH E1 β gene in the human, which diagnosis comprises determining the sequence of the nucleic acid at one or more of positions 457, 1191, 1198 and 1342 in the PDH E1 β gene as defined by the positions in SEQ ID NO: 1, and determining the status of the human by reference to polymorphism in the PDH E1 β gene; and
ii) administering an effective amount of a PDH drug.

15 9 Use of any one of the following in bioinformatic analysis:

i) any polymorphism defined in claim 1 or 2; or
ii) any haplotype defined in claim 3.

10 10 A use according to claim 9 comprising a bioinformatic analysis selected from
20 homology searching, mapping, haplotyping, genotyping or pharmacogenetic.